The diagnosis of early stage melanoma can be challenging histopathologically and can have a discordance rate as high as 27%. Non-Invasive gene expression testing for LINC and PRAME via a non-invasive pigmented lesion assay (PLA, 91% sensitivity, 69% specificity) has been validated against histopathology. Mutations in BRAF, NRAS and TERT promoter are found to correlate with melanoma tumor progression and histopathologic criteria.

We sought to validate the PLA against key driver mutations in melanoma. A prospective/retrospective analysis of 103 PLA adhesive patch samples, with consensus panel confirmed histopathologic diagnoses, were analyzed for hotspot mutations in BRAF (non-V600E), NRAS, and TERT. Furthermore, a prospective analysis of mutation frequency in 523 real-world PLA samples was performed.

97% percent of histopathologically confirmed melanoma samples were either PLA positive or mutation positive. Statistically significant differences in mutation frequency were observed between mel(+)/PLA+ and mel(-)/PLA(-) samples for hotspot mutations (75% vs. 15%, p<<0.0001). Mutations in adhesive patch samples were concordant with mutations in FFPE tissue blocks. TERT promoter mutations were the most prevalent (79%). Real-world PLA results showed that 89% of PLA(-) results were mutation negative, while 60% of PLA(+) results were mutation positive. There was no statistical difference in mutation frequency between validation samples and real-world samples.

This study confirms the high performance of the PLA. PLA positive tests identify high-risk lesions with driver mutations, while PLA negative test do not harbor these mutations. Gene expression and mutation analyses may enhance pigmented lesion assessment.

DermTech is the global leader in molecular dermatology, bringing precision medicine to the diagnosis and treatment of skin disease. We market and develop products that facilitate the early detection of skin cancers, assess inflammatory diseases, and customize drug treatments. DermTech analyzes skin biopsy samples collected non-invasively using an adhesive patch rather than a scalpel. Our mission is to transform dermatology by delivering highly accurate and objective information to the clinician to improve care and reduce costs and to pharma partners to support the development of targeted therapeutics. For additional information visit dermtech.com.